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(54) Title: HAPLOTYPES AND POLYMORPHISMS LINKED TO HUMAN THIOPURINE S-METHYLTRANSFERASE DEFICIENCIES

(57) Abstract: Haplotypes and polymorphisms of thiopurine S-methyltransferase (TPMT) are described that are linked to TPMT deficiencies which can cause potentially fatal toxicity when patients are treated with thiopurines like mercaptopurine, azathioprine, or thioguanine. The mutant alleles as well as PCR fragments, kits and methods for assaying the TPMT genotype of individual patients are disclosed. Furthermore, algorithms are disclosed that combine the genotypes of a set of single nucleotide polymorphisms to haplotypes that give a distinct information about the TPMT phenotype.